Reviewer's report

Title: Novel PTRF-CAVIN mutation in a child with mild myopathy and very mild congenital lipodystrophy

Version: 4 Date: 25 March 2013

Reviewer: Marwan Shinawi

Reviewer's report:

The manuscript is well written and the presentation of data is logical and convincing. However, I have the following minor comments:

1. I recommend to cite a recent article by Murakami et al. (PMID: 23489663) and discuss overlapping clinical findings and differences.

2. The official name of the gene is PTRF

3. Second paragraph p. 4: the first sentence should be reworded. One suggestion: “The patient was born, after an uncomplicated pregnancy, with APGAR scores of 8 at 1 minute and 9 at 5 minutes. He was noticed to have supinated left foot varus, mild dysmorphic features and (reduced motility?) (muscle weakness?hypotonia?)”

4. Last line p.6: “correct stop codon” should be changed to “original stop codon”.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests